

Case Report

Arnold-Chiari Type II Malformation: A Case Report and Review of Prenatal Sonographic Findings

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Abstract

The Arnold-Chiari malformation is a congenital abnormality of CNS, characterized by downward displacement the parts of the cerebellum, fourth ventricle, pons and medulla oblongata into the spinal canal. This malformation is one of causative factor of death in neonates and infants. A thorough understanding of the direct and indirect sonographic findings is necessary for diagnosis of Chiari II malformation in the developing fetus.

In this case report, we present a Chiari malformation II detected at 23 weeks of gestation by routinely sonographic screening. The Role of prenatal sonography in recognition of the malformation and prognostic value of these features are discussed.

Keywords: Arnold Chiari Malformation Type II, Prenatal Diagnosis, Ultrasonographic Screening

Introduction

Arnold-Chiari malformation with an incidence of 0.4:1000 live-birth is one of the CNS abnormalities that has formed 3% of all abortion and 1-2% recurrent risk (1) and is classified to three types. Type I consists of inferior displacement of the tonsils and cerebellum without displacement of the fourth ventricle or medulla. Arnold-Chiari malformation type II is the most common and seen in neonates and infants which characterized by displacement of cerebellar tonsils, parts of the cerebella fourth ventricle, pons and medulla oblongata through the foramen magnum into the spinal canal. This is usually associated with hydrocephalus and myelomeningocele (2) (Fig 1).

Chiari III malformation is a high cervical encephalomeningocele in which the medulla, fourth ventricle, and virtually the entire cerebellum reside (3).

The obvious sonographic findings are the frontal bone scalloping (lemon sign) and absent cerebellum or abnormal anterior curvature of the cerebellar hemispheres (banana sign) (4).

We report one case of Chiari malformation II detected at 23 weeks of gestation in routinely sonographic monitoring and describe direct and indirect sonographic findings that are the basis for diagnosis of Chiari II malformation.

Case Report

A 32-year-old pregnant woman, gravida 2, para 0, abortion 1, with 13 years of primary infertility, and no history of familial genetic disorders was admitted to Royan Institute (Infertility Clinic & Reproductive

Biomedicine). She had previous history of laparoscopy, hysteroscopy, ovarian cauterization, and polyp extraction.

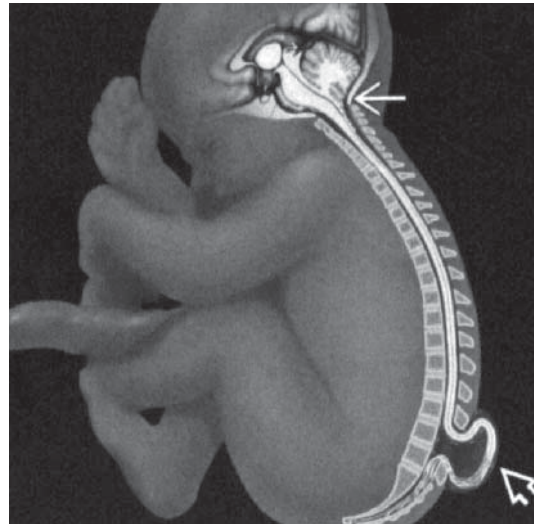


Fig 1: Sagittal graphic of Chiari II malformation with myelomeningocele (open arrow). Hindbrain herniation is the hallmark finding. The cerebellum is herniated (arrow) and the cisterna magna is obliterated.

The infertility factors included anovulation and teratospermia (normal morphology of 10%). After preliminary investigation, she underwent intrauterine insemination (IUI) twice with clomiphene citrate/ HMG

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protocol. The result of first cycle was negative for pregnancy. In the second treatment cycle, pregnancy occurred and a single gestational sac with alive fetus was seen at 7 weeks.

Sonographic examination at 13 and 15 weeks of gestation confirmed normal fetal anatomy. In routine triple test at 15 weeks, an elevated α -Fetoprotein (178mmol) was reported. Between 15 to 23 weeks of pregnancy, the patient had no workup in our center due to individual problem, unfortunately. In sonographic monitoring at 23 weeks of gestation, multiple fetal anomalies including microcephaly, lemon sign, mild ventriculomegaly, spinal bifida with myelomeningocele (27mm) (Fig 2), small cerebellum with downward displacement of medulla, fourth ventricle, cerebellum into the cervical spinal canal (Fig 3), obliteration of cisterna magna and club foot (Rocker-bottom feet type) were detected. According to these sonographic findings, the Arnold-Chiari malformation (type II) was confirmed and termination of pregnancy performed at 25 weeks of gestation.

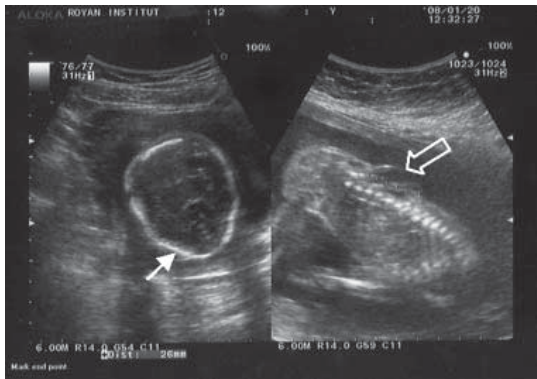


Fig 2: The lemon sign (arrowed) and myelomeningocele (open arrowed)



Fig 3: Downward displacement of the posterior fossa into the cervical spine

Discussion

Multiple studies have evaluated the accuracy of sonography for diagnosis of Chiari malformation (5-7). The feature of the Chiari II malformation that have been most useful are the infratentorial findings, these include

effacement of the cisterna magna (5) and deformation of the cerebellum which is called banana sign, although other infratentorial abnormalities are commonly observed postnatally (6-7).

Although the dominant feature of chiari malformation relate to the hindbrain, many supratentorial abnormalities have also been described (6-7). Included in these are callosal dysgenesis, a small third ventricle, enlarged interthalamic adhesions, a beaked tectum, polymicrogyria, heterotopias, skull deformities (the "lemon sign"), colpocephaly, and other cause of ventriculomegaly. Important among these is ventriculomegaly because visualization of the lateral ventricle is required on all routine sonography. Unfortunately, ventriculomegaly is considerably less common before 24 weeks than after 24 weeks in fetuses affected with myelomeningocele (8). The severity of posterior fossa (PF) deformity was graded to mild, moderate and severe. The PF deformity was considered mild when smaller than normal (<2mm) but identifiable; cisterna magna was present and the cerebella which was large enough to be easily identified, did not appear misshapen (Fig 4).

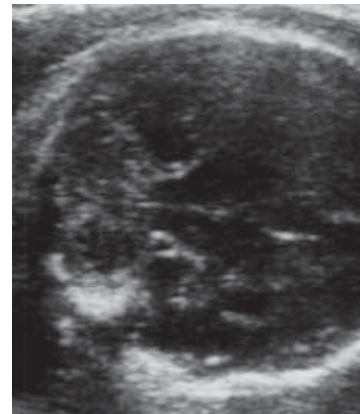


Fig 4: Mild PF deformity. Sonography depicts cisterna magna is smaller than normal (1mm) in size. The shape of cerebellum is normal.

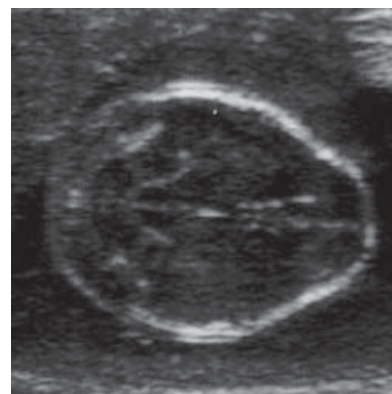


Fig 5: Moderate PF deformity. Sonograms show that the PF is somewhat small and the cisterna magna is affected. Cerebellar tissue can be confidently identified, although it demonstrates an abnormal contour (banana shape).

A moderate deformity was diagnosed when the PF subjectively appeared somewhat small, the cisterna magna was effaced and misshapen cerebellar tissue could confidently be identified (banana shape) (Fig 5).

The PF deformity was considered severe when PF to be very small, the cisterna magna was affected and little or no identifiable cerebellar tissue was visible (8) (Fig 6).

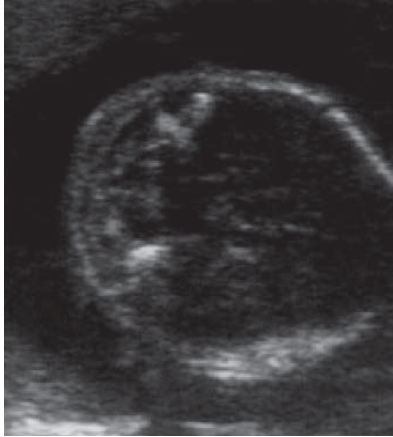


Fig 6: Severe PF deformity. The PF is very small, the cisterna magna is completely affected, and there is little identifiable cerebellar tissue.

According to this grading system, our case have moderate to severe PF deformity.

The cranial findings associated with the Chiari II malformation are found exclusively in fetuses with myelomeningocele. Therefore, identification of features of the Chiari II malformation virtually ensures that myelomeningocele is present. Probably, among these supratentorial findings are the so called lemon sign (inward scalloping of the frontal bones) and ventriculomegaly.

However, the lemon sign is frequently not present in later pregnancies (9) and can be seen in healthy fetuses (9, 10) and in other conditions (10). In addition, ventriculomegaly may be absent particularly before 24 weeks (8) but when is present, is nonspecific for myelomeningocele (11).

McLone and knepper (12) suggest that the VM seen with myelomeningoceles is secondary to the small size of the PF, which results in obstruction to the flow of cerebrospinal fluid at the level of the dysplastic tentorium, the aqueduct, the outlets of the fourth ventricle, or the foramen magnum.

If this theory is correct, we would expect to see some morphologic evidence demonstrated by neurosonograms of the fetus. It is reasonable to postulate that initial quantity of leaked cerebrospinal fluid would be variable and that the resulting effect on the PF would also be variable, resulting in a spectrum of PF malformations at prenatal neurosonography. Descriptors in the literature ranging from "effacement of the fetal cisterna magna" (5) to the banana-shaped cerebellum (13) to the absent cerebellum (14) have implied a continuum of severity of PF deformity.

The prevalence of VM in Bobcook study was much more common and often marked in fetuses with moderate or severe PF abnormalities, especially later in gestation (8). Although these data suggest that the qualitative appearance of the PF in the first half of gestation is predictive of the degree of VM likely to develop later.

Future studies will be helpful for further establishing that there is a causal relationship between the morphologic characteristics of the PF and the prevalence and severity of VM; for determining whether prenatal observations of the severity of PF deformities can be used to better predict outcome (8).

The diagnosis of myelomeningocele in a fetus is important for many reasons. It provides the parents with an opportunity to consider pregnancy termination. Among parents electing to continue the pregnancy, adequate counseling and psychological preparation can be provided (15).

In our patient, more accurate evaluation on spinal canal was performed after detection of "Lemon sign" and mild ventriculomegaly at 23 weeks of pregnancy.

In conclusion, the ultrasonographic prenatal screening is emphasized as the primary method of assessment of the early fetal malformation. Early diagnosis of such malformation helps to make decision to offer further fetal karyotyping or termination of pregnancy.

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